Search for the best indicators for the presence of a VPS13B gene mutation and confirmation of diagnostic criteria in a series of 34 patients genotyped for suspected Cohen syndrome

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Résumé en anglais	BACKGROUND: Cohen syndrome is a rare autosomal recessive inherited disorder that results from mutations of the VPS13B gene. Clinical features consist of a combination of mental retardation, facial dysmorphism, postnatal microcephaly, truncal obesity, slender extremities, joint hyperextensibility, myopia, progressive chorioretinal dystrophy, and intermittent neutropenia.PATIENTS AND METHODS: The aim of the study was to determine which of the above clinical features were the best indicators for the presence of VPS13B gene mutations in a series of 34 patients with suspected Cohen syndrome referred for molecular analysis of VPS13B. RESULTS: 14 VPS13B gene mutations were identified in 12 patients, and no mutation was found in 22 patients. The presence of chorioretinal dystrophy (92% vs 32%, p=0.0023), intermittent neutropenia (92% vs 5%, p<0.001), and postnatal microcephaly (100% vs 48%, p=0.0045) was significantly higher in the group of patients with a VPS13B gene mutation compared to the group of patients without a mutation. All patients with VPS13B mutations had chorioretinal dystrophy and/or intermittent neutropenia. The Kolehmainen diagnostic criteria provided 100% sensibility and 77% specificity when applied to this series. CONCLUSION: From this study and a review of more than 160 genotyped cases from the literature, it is concluded that, given the large size of the gene, VPS13B screening is not indicated in the absence of chorioretinal dystrophy or neutropenia in patients aged over 5 years. The follow-up of young patients could be a satisfactory alternative unless there are some reproductive issues.
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